



PRF1 gene

perforin 1

Normal Function

The *PRF1* gene provides instructions for making a protein called perforin. This protein is found in immune cells (lymphocytes) called T cells and natural killer (NK) cells, which destroy other cells. Perforin is involved in the process of cell destruction (cytolysis) and the regulation of the immune system.

Perforin is a major component of structures called cytolytic granules within T cells and NK cells. One of the main ways in which T cells and NK cells destroy other cells is to transport and secrete these cytolytic granules, which contain cell-killing proteins, onto the membranes of the target cells. Perforin helps create a channel through the membrane, allowing cytolytic proteins to enter the cell and trigger it to self-destruct.

This cytolytic mechanism also helps regulate the immune system by destroying unneeded T cells. Controlling the number of T cells prevents the overproduction of immune proteins called cytokines that lead to inflammation and which, in excess, cause tissue damage.

Health Conditions Related to Genetic Changes

familial hemophagocytic lymphohistiocytosis

More than 90 *PRF1* gene mutations have been identified in people with familial hemophagocytic lymphohistiocytosis. These mutations result in the production of a defective perforin protein or prevent the production of perforin. The resulting shortage of functional perforin prevents it from carrying out its role in cell destruction and immune system regulation, leading to the exaggerated immune response characteristic of familial hemophagocytic lymphohistiocytosis.

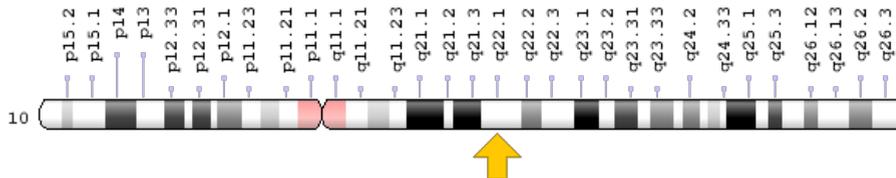
cancers

People with *PRF1* gene mutations are at increased risk of developing cancers of blood-forming cells (leukemia and lymphoma). Some of these individuals also have familial hemophagocytic lymphohistiocytosis. *PRF1* gene mutations impair the immune system's ability to destroy abnormal cells, allowing them to grow and divide in an uncontrolled way and leading to the development of cancer.

Chromosomal Location

Cytogenetic Location: 10q22.1, which is the long (q) arm of chromosome 10 at position 22.1

Molecular Location: base pairs 70,597,348 to 70,602,775 on chromosome 10 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- cytolysin
- FLH2
- HPLH2
- lymphocyte pore-forming protein
- lymphocyte pore forming protein
- MGC65093
- OTTHUMP00000019759
- P1
- PERF_HUMAN
- perforin-1
- perforin 1 (pore forming protein)
- perforin-1 precursor
- PFN1
- PFP

Additional Information & Resources

Educational Resources

- Immunology (5th edition, 2001): Cytotoxic effector proteins that trigger apoptosis are contained in the granules of CD8 cytotoxic T cells
<https://www.ncbi.nlm.nih.gov/books/NBK27101/#A1080>

GeneReviews

- Hemophagocytic Lymphohistiocytosis, Familial
<https://www.ncbi.nlm.nih.gov/books/NBK1444>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28PRF1%5BTIAB%5D%29+OR+%28perforin+1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- PERFORIN 1
<http://omim.org/entry/170280>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_PRF1.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=PRF1%5Bgene%5D>
- HGNC Gene Family: C2 domain containing
<http://www.genenames.org/cgi-bin/genefamilies/set/823>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=9360
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/5551>
- UniProt
<http://www.uniprot.org/uniprot/P14222>

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